



CRGM DOLJ



UMF CRAIOVA

## CURS PRACTIC DE GENETICĂ MOLECULARĂ

”Tehnici moleculare în diagnosticul și screening-ul genetic,  
de la nucleotid la genom”

Craiova, 3-5 SEPTEMBRIE 2017

	<i><b>DUMINICĂ, 3 SEPTEMBERIE, 2017</b></i>
11:00 – 12:30	CAZARE & INREGISTRARE PARTICIPANTI
12:30 – 13:30	PRÂNZ
<b>13:30 – 14:00</b>	<b>DESCHIDEREA OFICIALĂ A CURSULUI</b> Mihai Ioana
<b>14:00 – 14:30</b>	<b>CITOGENETICĂ CONVENȚIONALĂ</b> Florin Burada
<b>14:30 – 15:00</b>	<b>FISH</b> Vlad Gorduza
15:00 – 15:15	Pauză de cafea
<b>15:15 – 16:00</b>	<b>FLUXURILE DE LUCRU ÎN LABORATORUL DE GENETICĂ MOLECULARĂ</b> Alina Cimpoeru, Mihai Cucu, Razvan Plesea, Mihai Ioana
<b>16:00 – 16:40</b>	<b>REAL TIME PCR – TEHNICA &amp; INTERPRETARE REZULTATE</b> Alina Cimpoeru, Elena Popescu
16:40 – 16:55	<b>PREZENTARE ROCHE EXTRACTOR AUTOMAT</b> Roche
<b>16:55 – 17:55</b>	<b>“LAB IN A TUBE” - TRAINING PRACTIC PE INSTRUMENTUL COBAS LIAT</b> Roche
17:55 – 18:00	<b>SINTEZA</b> Mihai Ioana



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<i><b>LUNI, 4 SEPTEMBERIE, 2017</b></i>	
<b>09:00 – 09:20</b>	<b>ANALIZA DE FRAGMENTE</b> Mihai Ioana
<b>09:20 – 09:40</b>	<b>QF-PCR – PROTOCOL TEHNICA</b> Anca Riza
<b>09:40 – 10:40</b>	<b>QF-PCR - APLICATII</b> Anca Riza, Adela Ciocoiu, Stefania Dorobantu
10:40 – 11:10	PAUZĂ DE CAFEA
<b>11:10 – 11:30</b>	<b>MLPA – TEHNICA</b> Razvan Plesea
<b>11:30 – 13:30</b>	<b>MLPA – PROTOCOL PRACTIC, PREGATIRE APARAT, ANALIZA REZULTATE, DISCUTII</b> Razvan Plesea, Alina Cimpoeru, Elena Popescu, Mihai Cucu
13:30 – 14:30	PRÂNZ
<b>14:30 – 16:00</b>	<b>ACGH – PROTOCOL, APLICATII, DISCUTII</b> Simona Serban Șoșoi, Andrei Pirvu, Ioana Streata
16:00 – 16:30	PAUZĂ DE CAFEA
<b>16:30 – 18:00</b>	<b>GWAS</b> Anca Riza
18:00 – 18:15	<b>SINTEZA</b> Mihai Ioana



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<i><b>MARȚI, 5 SEPTEMBERIE, 2017</b></i>	
<b>09:00 – 10:00</b>	<b>DE CE NGS?</b> Ioana Streata, Mihai Ioana
<b>10:00 – 11:00</b>	<b>UTILIZAREA TEHNICILOR DE SECVENȚIERE DE ULTIMĂ GENERAȚIE (NGS) ÎN IDENTIFICAREA VARIANTELOR NUCLEOTIDICE SI STRUCTURALE DIN GENOMUL UMAN</b> Horia Stanescu
11:00 – 11:30	PAUZĂ DE CAFEA
<b>11:30 – 13:00</b>	<b>Generare rezultate prin metodele NGS</b> Razvan Plesea, Alina Cimpoeru, Elena Popescu, Mihai Cucu
13:00 – 14:00	PRÂNZ
<b>14:00 – 15:30</b>	<b>WORKSHOP INTERPRETARE REZULTATE NGS – I</b> Horia Stanescu
15:30 – 16:00	PAUZĂ DE CAFEA
<b>16:00 – 17:30</b>	<b>WORKSHOP INTERPRETARE REZULTATE NGS – II</b> Horia Stanescu
17:30 – 18:00	<b>INCHIDEREA OFICIALĂ A CURSULUI</b> Mihai Ioana

**The Course is credited with 18 EMC points**



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## CONFERENCE OF THE ROMANIAN SOCIETY OF MEDICAL GENETICS SEPTEMBER 6 – 8, 2017 CRAIOVA

The time allocated to each presentation includes 3 - 5 minutes for questions, answers and comments. We advise each speaker to respect the time allocated.

**WEDNESDAY, September 6, 2017**

**10:00 – 11:00 REGISTRATION**

**11:00 – 11:30 OFFICIAL OPENING OF THE CONFERENCE**

<b>11:30 – 13:00</b>	<b>PLENARY SESSION 1</b> <i>Moderators: MARIA PUIU, MIHAI IOANA</i>
<b>11.30 -12.00</b>	<b>SP1.1. MEDICAL PRACTICE IN THE GENOMIC ERA</b> Bembea Marius, Jurca Claudia, Kozma Kinga, Iuhas Oana, Petcheși Codruța
<b>12.00-12.30</b>	<b>SP1.2. GRANT PROPOSALS – STRATEGIES AND TACTICS</b> Mihai Niculescu
<b>12.30 -12.55</b>	<b>SP1.3. INTRODUCTION TO INBORN ERRORS OF METABOLISM</b> Natalia Usurelu
<b>12.55-13.00</b>	<b>Discutii</b>

**13.00 -14.30 LUNCH BREAK**

<b>14.30 -16.15</b>	<b>PLENARY SESSION 2</b> <i>Moderators: CRISTINA RUSU, AMELIA DOBRESCU</i>
<b>14.30 - 14.50</b>	<b>SP2.1. CENTER FOR GENOMIC MEDICINE TIMISOARA-SHORT DESCRIPTION, COLLABORATIONS AND RESULTS</b> Maria Puiu, Nicoleta Andreescu, Adela Chirita Emandi, Simona Farcas



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14.50 - 15.10	<b>SP2.2. HINT: HYPERURICAEMIA INDUCED INFLAMMATION: TARGETING THE CENTRAL ROLE OF URIC ACID IN RHEUMATOLOGIC AND CARDIOVASCULAR DISEASE</b> Tania O. Crișan, Maartje C.P. Cleophas, Cristian G. Andra, Alex Bolunduț, Marius F. Farcaș, Ioana Hotea, Cristina Pamfil, Simona Rednic, Ioan V. Pop, Radu A. Popp, Mihai G. Netea, Leo A.B. Joosten
15.10-15.30	<b>SP2.3. FUSE: FUNCTIONAL GENOMICS IN SEVERE INFECTIONS</b> Anca Riza, Ioana Streata, Mihai Ioana, Mihai G. Netea
15.30-15.50	<b>SP2.4. NUTRIGEN: DEFINING GENETIC SIGNATURES TO PREDICT THE EFFICACY OF DISEASE MANAGEMENT WITH MEDICAL FOODS IN INDIVIDUALS WITH OBESITY, DYSLIPIDEMIAS AND INSULIN RESISTANCE</b> Maria Puiu, Nicoleta Andreescu, Adela Chirita-Emandi, , Mihai D Niculescu
15.50-16.20	<b>SP2.5. ROCHE SYMPOSIUM</b>
16.20-16.30	<i>Discutii</i>

**16.30 -17.00 COFFEE BREAK, POSTER & EXHIBITION VIEWING**

17.00 – 19.00	<b>PLENARY SESSION 3</b> <i>Moderators: MONICA ZELENIUC, FLORIN BURADA</i>
17.00 - 17.15	<b>SP3.1. miRNA POLYMORPHISMS AND RISK OF GASTRIC CANCER IN A ROMANIAN POPULATION</b> Burada Florin, Cîmpeanu Raluca Alexandra, Cucu Mihai Gabriel, Ioana Mihai, Rogoveanu Ion
17.15 - 17.30	<b>SP3.2. TNF-A 238A&gt;G GENE POLYMORPHISMS ARE NOT ASSOCIATED WITH FLT3 ITD GENE MUTATION AND THE RISK OF DEVELOPING AML</b> Andrei Crauciuc, Florin Tripon, Valeriu Moldovan, Bogliș Alina, Ioan Macarie, Claudia Bănescu
17.30 - 17.45	<b>SP3.3. ANALYSIS OF RISK FACTORS FOR THE OCCURRENCE AND EVOLUTION OF CHRONIC MYELOID LEUKEMIA</b> Andra-Maria Tomescu, Mihaela Dragomir, Silvia Aposteanu, Rodica Talmaci, Dănuț Cimponeriu, Ileana Stoica
17.45 – 18.00	<b>SP3.4. LIQUID BIOPSY: VALUABLE TOOL IN DIAGNOSIS AND MONITORING OF PERSONALIZED THERAPY IN LUNG CANCER</b> Cardos Georgeta, Motoc Rozalia, Bucur Gabriela Georgeta, Vasile Daniela, Iorga Polixenia, Apostol Pompilia Petruta
18.00 – 18.15	<b>SP3.5. FAMILY HISTORY – ESSENTIAL TOOL FOR DIAGNOSIS AND CANCER RISK ASSESSMENT</b>



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	Caba Lavinia, Ciofu Mihai Liviu, Haba Danisia, Burlea Marin, Păduraru Gabriela, Pânzaru Monica, Butnariu Lăcrămioara, Resmeriță Irina, Popescu Roxana, Gorduza Eusebiu Vlad
18.15-18.30	<b>SP3.6. INTERPRETATION AND CLINICAL VALIDATION OF BRCA1 AND BRCA2 SEQUENCING RESULTS IN BREAST CANCER - THE NEED FOR INTEGRATED GENETIC DATABASES</b> Viorica Radoi, Roxana Bohiltea, Gratiela Chelu, Cristina Dragomir, Bianca Basangiu, Oana Istrate, Sandra Grigore, Marinela Malageanu, Antoanela Curici, Radu Ursu
18.30 – 18.40	<b>DISCUSSIONS</b>

19.30 -22.00 DINNER

THURSDAY, 7 September, 2017

09.00 – 10.30	<b>PLENARY SESSION 4</b> <i>Moderators: SIMONA FARCAȘ, HORIA STĂNESCU</i>
09.00 - 09.15	<b>SP4.1. MASSIVE LACTIC ACIDOSIS IN ROMA ETHNICITY - CONSIDER TMEM70 GENE MUTATION</b> Harapu Iulia, Boia Mariana, Manea Aniko, Popoiu Anca, Puiu Maria, Markéta Tesařová, Martin Magner, Chirita-Emandi Adela
09.15 - 09.30	<b>SP4.2. MOLECULAR-GENETIC DIAGNOSIS IN WILSON'S MOLDOVAN PATIENTS</b> Victoria Hlistun, Andree Zibert, Victoria Sacara
09.30 - 09.45	<b>SP4.3. ALPHA-1 ANTITRYPSIN GENE VARIATIONS AND GOUTY INFLAMMATION</b> Alexandru C. Bolunduț, Cristian G. Andra, Marius F. Farcaș, Ioana Hotea, Cristina Pamfil, Simona Rednic, Leo A.B. Joosten, Ioan V. Pop, Radu A. Popp, Tania O. Crișan
09.45 - 10.00	<b>SP4.4. GENETIC RISK VARIANTS ASSOCIATED WITH DIABETES MELLITUS IN A COHORT OF ROMANIAN MEN</b> R.I Ursu, P. Iordache, V. Radoi, N. Cucu, G.F. Ursu, R. Bohiltea, I. Craciunescu, C. Staicu, V. Calota, A. Voinoiu, I. Csiki, D. Mates, E. Poenaru, C.L. Bohiltea, A. Manolesc, V. Jinga.
10.00-10.15	<b>SP4.5. GENETIC AND EPIGENETIC VARIATION IN GOUT AND HYPERURICAEMIA</b> Tania O. Crișan, Maartje C.P. Cleophas, Cristian G. Andra, Alex Bolunduț, Marius F. Farcaș, Ioana Hotea, Cristina Pamfil, Simona Rednic, Ioan V. Pop, Radu A. Popp, Mihai G. Netea, Leo A.B. Joosten



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<b>10.15-10.30</b>	<b>SP4.6. THE VARIABILITY OF SCREENING CRITERIA IN CONGENITAL DISORDERS OF GLYCOSYLATION</b> Blaniță D., Sacară V., Boiciuc C., Hlistun V., Grosu I., Dorif A., Morava E., Lefeber D., Wevers R., Uşurelu N.
<b>10.30-11.00</b>	<b>SP4.7. ELTA 90 MR SYMPOSIUM</b> <b>Teodor Zamfirov</b>

**11.00 -11.30 COFFEE BREAK, POSTER & EXHIBITION VIEW**

<b>11.30 – 13.30</b>	<b>PLENARY SESSION 5</b> <i>Moderators: MIHAELA BARDOȘ, MARIUS BEMBEA</i>
<b>11.30 – 12.15</b>	<b>SP5.1. PARTNERSHIP BETWEEN MINISTRY OF HEALTH, MEDICAL GENETICS REGIONAL CENTRES AND SRGM - PRESENT AND PERSPECTIVES</b> Mihaela Bardos, Lidia Onofrei, Dan Dumitru
<b>12.15 - 12 .25</b>	<b>SP5.2. CRGM TIMISOARA ACTIVITY</b> Maria Puiu
<b>12.25 - 12.35</b>	<b>SP5.3. CRGM CLUJ ACTIVITY</b> Camelia Al-Khzouz
<b>12.35 - 12.45</b>	<b>SP5.4. CRGM ORADEA ACTIVITY</b> Claudia Jurca
<b>12.45 – 12:55</b>	<b>SP5.5. CRGM BUCURESTI ACTIVITY</b> Vasilica Plaiasu
<b>12.55 – 13:05</b>	<b>SP5.6. CRGM IASI ACTIVITY</b> Cristina Rusu
<b>13.05 – 13:15</b>	<b>SP5.7. CRGM DOLJ ACTIVITY</b> Mihai Ioana
<b>13.15 – 13.25</b>	<b>SP5.8. NATIONAL INSTITUTE OF FORENSICS “MINA MINOVICI” – GENETICS LABORATORY ACTIVITY</b> Ligia Barbarii, Elena Neagu

**13.30 -14.30 LUNCH BREAK**

<b>14.30 – 16.20</b>	<b>PLENARY SESSION 6</b> <i>Moderators: EMILIA SEVERIN, DORICA DAN</i>
<b>14.30 - 14 .50</b>	<b>SP6.1. SOCIAL IMPACT OF RARE DISEASES</b> Dorica Dan, Zsuzsa Almasi, Florina Breban, Maria Acaralitei, Dorisz Veres, Alexandra Toporan



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14.50 - 15.10	<b>SP6.2. O PERSPECTIVA ASUPRA NEVOILOR DE DEZVOLTARE A ASISTENTEI PACIENTULUI CU BOLI RARE</b> Izabela Tudorache
15.10 - 15.20	<b>SP6.3. ASOCIATIA OAMENILOR MICI ROMANIA</b> Alina Tatuca
15:20 – 15:50	<b>SP6.4. GENZYME SYMPOSIUM</b>

**15.50 -16.15 COFFEE BREAK, POSTER & EXHIBITION VIEWING**

16.15 – 18.30	<b>PLENARY SESSION 7</b> <i>Moderators: VIORICA RĂDOI, MIHAI NICULESCU</i>
16.15 - 16 .35	<b>SP7.1. FREQUENT FORMS OF SUBTELOMERIC REARRANGEMENTS – EVOCATIVE FEATURES THAT FACILITATE THEIR RECOGNITION</b> Cristina Rusu, Monica Pânzaru, Lăcrămioara Butnariu, Mihaela Grănescu, Eusebiu Vlad Gorduza, Adriana Sireteanu, Roxana Popescu
16.35 - 16.50	<b>SP7.2. THE IMPORTANCE OF CYTOGENETIC AND MOLECULAR ANALYSES IN ELUCIDATION OF ETIOLOGY OF PLURIMALFORMATIVE SYNDROMES</b> Mihaela Grănescu, Lavinia Caba, Monica Pânzaru, Lăcrămioara Butnariu, Elena Braha, Roxana Popescu, Setalia Popa, Cristina Rusu, Georgeta Cardos, Monica Zeleniuc, Violeta Martiniuc, Eusebiu Vlad Gorduza
16.50 - 17.05	<b>SP7.3. EVALUATION BY ARRAYCGH OF ROMANIAN PATIENTS WITH DEVELOPMENTAL DISABILITY AND CONGENITAL ANOMALIES</b> Monica Zeleniuc, Petruta Gurban, Gabriel Dinulescu, Vasilica Plaiasu, Vlad Gorduza, Carmen C Diaconu, Diana Ozunu, Gabriela Motei, Setalia Popa, Georgeta Cardos
17.05-17.20	<b>SP7.4. THE IMPLICATION OF CNVS IN PATIENTS WITH OBESITY ASSOCIATED WITH INTELLECTUAL DISABILITY</b> Osan S, Miclea D, Popp RA, Puiu M, Bucerzan S, Cret V, Crisan M, Lazea C, Farcaș M, Andreescu N, Chirita-Emandi A, AlKhzouz C
17.20-17.35	<b>SP7.5. GENETIC DIAGNOSTIC THROUGH aCGH IN GLOBAL DEVELOPMENT DELAY ASSOCIATED WITH CONGENITAL ANOMALIES – CRGM DOLJ UPDATED ACTIVITY</b> Ioana Streață, Andrei Pirvu, Simona Șerban-Șosoi, Burada F., Ioana M.
17.35-17.50	<b>SP7.6. NEW FEATURES OF ORPHANET IN 2017</b> Cristina Rusu, Monica Pânzaru
17.50 – 18.05	<b>SP7.7. SRGM ACTIVITY IN THE PAST YEAR</b> Maria Puiu, Adela Chiriță
18.05 – 18.20	<b>DISCUSSIONS</b>





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19.30-22.00 DINNER

Friday, 8 September, 2017

09.00 – 10.45	<b>PLENARY SESSION 8</b> <i>Moderators: VLAD GORDUZA, FRANCISC MIXICH</i>
09.00 - 09 .15	<b>SP8.1. PRENATAL SCREENING AND DIAGNOSIS IN CHROMOSOMAL ANOMALIES: INSIGHTS FROM PAUCISYMPTOMATIC CASES.</b> Tudorache Stefania, Sorop-Florea Maria, Dragusin Roxana Cristina, Patru Ciprian Laurentiu, Iliescu Dominic Gabriel, Burada Florin, Ioana Mihai, Dobrescu Amelia, Cara Laura Monica
09.15 - 09.30	<b>SP8.2. NIPT (NON-INVASIVE PRENATAL TESTING) – A CHALLENGING SOLUTION</b> Dominic Iliescu, Stefania Tudorache
09.30 – 09.45	<b>SP8.3. PRENATAL GENETIC COUNSELING IN CONGENITAL ANOMALIES</b> Mihaela Amelia Dobrescu, Cristina Durac, Gratiela Chelu, Raluca Sanziana Poenaru, Carmen-Bianca Petre-Mandache , Stefania Tudorache
09.45 – 10.05	<b>SP8.4. TESTING THE IMPACT OF INTERACTIONS BETWEEN GENETIC AND NON-GENETIC RISK FACTORS ON FEMALE FERTILITY</b> Oana Apîrcioaie, Remus Nica, Silvia Nica, Spandole Dinu Sonia, Barbăroșie Catalina, Alexiu Andrada, Irina Radu, Mihai Toma, Cimponeriu Dănuț, Ileana Stoica
10.00 – 10:15	<b>SP8.5. QUANTIFICATION OF TREC/KREC LEVELS IN BLOOD OF NEWBORNS IN MOLDOVA</b> Dorif Alexandr, Gordukova Maria, Filipenko Maxim, Andries Lucia, Sacara Victoria
10:15 – 10:30	<b>SP8.6. THE COMPLEX MANAGEMENT OF A PRENATAL TESTING CASE FOR DYSTROPHIC EPIDERMOLYSIS BULLOSA</b> Cristina Gug, Adrian Ratiu, Caius Solovan
10.30 – 10.45	<b>Discutii</b>

11:00 – 11:30 POSTER AWARDS. CONCLUSIONS. OFFICIAL CLOSING OF THE CONFERENCE.



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# POSTER presentations

## **P1. FROM THEORY TO PRACTICE: A CONGENITAL HEART DISEASE AND AMBIGUOUS GENITALIA CASE AND LITERATURE REVIEW**

Mădălina Anciu<sup>1</sup>, Claudia Bănescu<sup>2,3</sup>, Delia Tătar<sup>1</sup>, Manuela Camelia Cucerea<sup>1,3</sup>, Alina Bogliș<sup>2,3</sup>

<sup>1</sup>Department of Neonatology, Emergency Clinical County Hospital of Țirgu Mureș, Romania

<sup>2</sup>Department of Genetics, Emergency Clinical County Hospital of Țirgu Mureș, Romania

<sup>3</sup>University of Medicine and Pharmacy Țirgu Mureș, Romania

## **P2. RAS MUTATION TESTING EXTENDED IN ROMANIAN PATIENTS WITH COLORECTAL CANCER (CRC)**

Apostol Pompilia<sup>1</sup>, Dinu Sorin<sup>1</sup>, Gurban Petruta<sup>1</sup>, Iordache Florin<sup>1</sup>, Schenker Michael<sup>2</sup>, Turcu Alina<sup>3</sup>, Bucur Gabriela<sup>1</sup>, Cardos Georgeta<sup>1</sup>

<sup>1</sup>Personal Genetics - Medical Genetics Center, Bucharest

<sup>2</sup>"Sfantul Nectarie" Center of Oncology, Craiova

<sup>3</sup>Municipal Hospital, Ploiesti

## **P3. MOSAIC 46XX / 45, X / 47, XXX IN A COUPLE WITH A HISTORY OF SPONTANEOUS MISCARRIAGE AND 4 FAILED IN VITRO FERTILIZATION TECHNIQUES**

Diter Atasi<sup>1</sup>, Bunescu Cosmina<sup>2</sup>

<sup>1</sup>Lucian Blaga University, Faculty of Medicine Sibiu

<sup>2</sup>Polisano Clinic

## **P4. THE PERICENTRIC INVERSION ON CHROMOSOME 9 (46, XY, INV (9)) (P11; Q13); (46, XX, INV (9)) (P11; Q13) IT'S ONE OF THE CAUSES OF COUPLE INFERTILITY**

Diter Atasi<sup>1</sup>, Bunescu Cosmina<sup>2</sup>

<sup>1</sup>Lucian Blaga University, Faculty of Medicine Sibiu

<sup>2</sup>Polisano Clinic

## **P5. NO ASSOCIATION BETWEEN COPY NUMBER CHANGES AND FLT3 OR DNMT3A GENE MUTATIONS IN ACUTE MYELOID LEUKEMIA**

Banescu Claudia<sup>1</sup>, Crauciuc Andrei<sup>2</sup>, Moldovan Valeriu<sup>1</sup>, Bogliș Alina<sup>2</sup>, Lazar Erzsebeth<sup>2</sup>, Tripon Florin<sup>2</sup>



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<sup>1</sup>Genetics Laboratory, Center for Advanced Medical and Pharmaceutical Research, University of Medicine and Pharmacy Tirgu Mures, Romania <sup>2</sup>University of Medicine and Pharmacy Tirgu Mures, Romania

## **P7. PROGNOSTIC VALUE OF ABCG2 GENE Q141K POLYMORPHISM IN ROMANIAN PATIENTS WITH ACUTE MYELOID LEUKEMIA WITH FLT3 MUTATION**

Bogliš Alina<sup>1</sup>, Tripon Florin<sup>1</sup>, Crauciuc Andrei<sup>1</sup>, Moldovan Valeriu<sup>2</sup>, Trifa Adrian<sup>3</sup>, Lazar Erzsebeth<sup>1</sup>, Bănescu Claudia<sup>1,2</sup>

<sup>1</sup>University of Medicine and Pharmacy Tirgu Mures, Romania

<sup>2</sup>Genetics Laboratory, Center for Advanced Medical and Pharmaceutical Research, University of Medicine and Pharmacy Tirgu Mures, Romania

<sup>3</sup>University of Medicine and Pharmacy “Iuliu Hatieganu” Cluj Napoca, Romania

## **P8. UTILITY OF GENETIC TESTING IN AUTISTIC SPECTRUM DISORDERS – PARENTS` PERSPECTIVES**

Roxana Elena Bohiltea<sup>1</sup>, Radu-Ioan Ursu<sup>2</sup>, Laurentiu Camil Bohiltea<sup>2</sup>, Viorica Elena Radoi<sup>2</sup>

<sup>1</sup>SUUB Department of Obstetrics and Gynecology, “Carol Davila” University of Medicine and Pharmacy, Bucharest

<sup>2</sup>Department of Medical Genetics, “Carol Davila” University of Medicine and Pharmacy, Bucharest

## **P9. CLINICAL AND ENDOCRINE FEATURES IN A PATIENT WITH THE 45,X/47,XYY KARYOTYPE**

Braha Elena Emanuela<sup>1</sup>, Popa Oana<sup>1</sup>, Nedelcu Ioana<sup>1</sup>, Poiană Cătălina<sup>1,2</sup>

<sup>1</sup>National Institute of Endocrinology ‘C.I. Parhon’, Bucharest

<sup>2</sup>UMF ‘Carol Davila’, University of Medicine and Pharmacy, Bucharest

## **P10. A REPORT OF A NEW CASE WITH DE NOVO 11 Q TERMINAL DELETION SYNDROME (JACOBSEN SYNDROME)**

Lacramioara Ionela Butnariu, Cristina Rusu, Monica Panzaru, Lavinia Caba, Roxana Popescu, Eusebiu Vlad Gorduza

University of Medicine and Pharmacy “Grigore T. Popa”, Department of Medical Genetics, Iasi, Romania

## **P11. PRENATAL DIAGNOSIS OF ALOBAR HOLOPROSENCEPHALY ASSOCIATED WITH CYCLOPIA**

Andreea Roxana Cioca<sup>1</sup>, Flavia Anne-Elise Szekely<sup>1</sup>, Mihai Narcis Morar<sup>1</sup>, Nicoleta Andreescu<sup>2</sup>, Maria Puiu<sup>1,2</sup>, Simona Farcaș<sup>2</sup>

<sup>1</sup>Department of Medical Genetics, “Louis Țurcanu” Emergency Hospital for Children, Timișoara, Romania

<sup>2</sup>Department of Genetics, “Victor Babeș” University of Medicine and Pharmacy, Timișoara, Romania



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## **P12. TESTING THE ASSOCIATION BETWEEN ACE ID AND AT1R A1166C POLYMORPHISMS AND DIABETES MELLITUS OR HYPERTENSION**

Anne Marie Crăciun<sup>1</sup>, Remus Nica<sup>2</sup>, Monica Muraru<sup>3</sup>, Mihai Toma<sup>2,3</sup>, Silvia Nica<sup>4</sup>, Cătălina Barbăroșie<sup>3</sup>, Andrada Alexiu<sup>3</sup>, Irina Radu<sup>3</sup>, Cimponeriu Dănuț<sup>3</sup>, Lavinia Berca<sup>5</sup>

<sup>1</sup>NIDNMD Nicolae Paulescu, Bucharest

<sup>2</sup>Dr. Carol Davila Central Military Emergency University Hospital

<sup>3</sup>Faculty of Biology, University of Bucharest

<sup>4</sup>Bucharest Emergency University Hospital

<sup>5</sup>National Institute of Research and Development for Food Bioresources, Romania

## **P13. RELEVANCE OF CYTOGENETICS ANALYSIS IN DIAGNOSIS AND MONITORING CHRONIC MYELOID LEUKEMIA PATIENTS**

Diana Georgiana Dobreă<sup>1</sup>, Georgiana Petrică-Matei<sup>1</sup>, Cristina Ionescu<sup>1</sup>, Raluca Manolache<sup>2</sup>, Carmen Saguna<sup>2</sup>, Oana Stanca<sup>3</sup>, **Monica Zeleniuc**<sup>1,4</sup>

<sup>1</sup>Personal Genetics - Medical Genetics Center, Bucharest, Romania

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Hrapşa Iona<sup>1</sup>, Farcaş Marius<sup>1</sup>, Oşan Sergiu<sup>1</sup>, Coroamă Constantin-Ionuţ<sup>1</sup>, Miclea Diana<sup>1,2</sup>, Popp Radu<sup>1</sup>, Al-Khzouz Camelia<sup>2</sup>

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Jurca Claudia<sup>1,2</sup>, Bembea Marius<sup>2</sup>, Kozma Kinga<sup>1,2</sup>, Petchesi Codruta<sup>1,2</sup>, Iuhas Oana<sup>2</sup>, Szilagyi Ariana<sup>1,2</sup>, Balmos Andreea<sup>1,2</sup>, Vesa Cosmin<sup>1</sup>, Jurca Alexandru<sup>1</sup>

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MARCHIAN SANDA

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Moldovan Valeriu<sup>1</sup>, Boglis Alina<sup>2</sup>, Crauciuc Andrei<sup>2</sup>, Tripon Florin<sup>2</sup>, Banescu Claudia<sup>1</sup>

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<sup>3</sup>Clinical Hospital Pantelimon Bucharest

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Remus Nica<sup>1</sup>, Silvia Nica<sup>2</sup>, Tomescu Andra<sup>3</sup>, Monica Muraru<sup>3</sup>, Alexiu Andrada<sup>3</sup>, Oana Apîrcioaică<sup>3</sup>, Radu Irina<sup>3</sup>, Toma Mihai<sup>1,3</sup>, Cimponeriu Danuț<sup>3</sup>, Berca Lavinia<sup>4</sup>

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Oana Păvăloaia<sup>2</sup>, Georgeta Orășanu<sup>4</sup>, Iuliana Stârcea<sup>3</sup>, Irina Resmeriță<sup>1,2</sup>, Roxana Popescu<sup>1,3</sup>, Monica Pânzaru<sup>1,3</sup>, Lăcrămioara Butnariu<sup>1,3</sup>, Mircea Covic<sup>1,3</sup>, Cristina Rusu<sup>1,3</sup>

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Perva Iulia<sup>1</sup>, Weiss Janneke<sup>2</sup>, Waisfisiz Quinten<sup>2</sup>, Chirita-Emandi Adela<sup>1</sup>, Zimbru Cristian<sup>1,3</sup>, Andreescu Nicoleta<sup>1</sup>, Puiu Maria<sup>1</sup>

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<sup>3</sup>Politehnica University, Timisoara

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Petre-Mandache Carmen-Bianca<sup>1</sup>, Serban-SoSoi Simona<sup>2</sup>, Popescu-Hobeanu Gabriela<sup>2</sup>, Cucu Mihai Gabriel<sup>2</sup>, Streata Ioana<sup>2</sup>, Dobrescu Mihaela-Amelia<sup>2</sup>, Ioana Mihai<sup>2</sup>, Sorin Dinescu<sup>2</sup>, Burada Florin<sup>2</sup>

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<sup>2</sup> UMF Craiova



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Georgiana Petrica-Matei<sup>1</sup>, Georgeta Cardos<sup>1</sup>, Roxana Bohiltea<sup>2</sup>, Roxana Constantin<sup>3</sup>, Ana-Maria Chiscop<sup>3</sup>, Alina Dascalu<sup>3</sup>, Diana Georgiana Dobrea<sup>1</sup>, Camil Laurentiu Bohiltea<sup>4</sup>, Monica Zeleniuc<sup>1,4</sup>

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PLESEA I.E. Razvan Mihail<sup>1, 2</sup>, POPESCU Elena Leocadia<sup>1,3</sup>, CUCU M. Mihai Gabriel<sup>1,2</sup>, CIMPOERU Alina Liliana<sup>1</sup>, IOANA Mihai<sup>1,2</sup>

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**Radu Cristina Georgiana**<sup>2</sup>, Tripon Florin<sup>2</sup>, Crauciuc George Andrei<sup>2</sup>, Bogliș Alina<sup>2</sup>, Demian Smaranda<sup>3</sup>, Bănescu Claudia<sup>1</sup>

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**Radu Cristina Georgiana**<sup>2</sup>, Tripon Florin<sup>2</sup>, Crauciuc George Andrei<sup>2</sup>, Bogliș Alina<sup>2</sup>, Demian Smaranda<sup>3</sup>, Bănescu Claudia<sup>1</sup>

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Irina Resmeriță<sup>1,2</sup>, Sebastian Cozma<sup>3</sup>, Roxana Popescu<sup>1,4</sup>, Monica Pânzaru<sup>1,4</sup>, Lavinia Caba<sup>1</sup>, Oana Păvăloaia<sup>2</sup>, Lăcrămioara Butnariu<sup>1,4</sup>, Cristina Rusu<sup>1,4</sup>

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Irina Savu<sup>1</sup>, Cristina Olariu<sup>2</sup>, Gabriela Doros<sup>2</sup>, Simona Farcaș<sup>1</sup>, Adela Chirita Emandi<sup>1</sup>, Maria Puiu<sup>1</sup>

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Flavia Anne-Elise Szekely<sup>1</sup>, Andreea Roxana Cioca<sup>1</sup>, Nicoleta Andreescu<sup>2</sup>, Maria Puiu<sup>1, 2</sup>, Simona Farcaș<sup>2</sup>

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Tripon Florin<sup>1</sup>, Crauciuc George Andrei<sup>1</sup>, Bogliș Alina<sup>1</sup>, Moldovan Valeriu<sup>1</sup>, Duicu Carmen<sup>1</sup>, Lazar Erzsebeth<sup>1</sup>, Trifa Pavel Adrian<sup>2</sup>, Bănescu Claudia<sup>1, 3</sup>

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Turcan Doina<sup>1</sup>, Grosu Iulia<sup>1</sup>, Sacară Victoria<sup>1</sup>

<sup>1</sup>Institute of Mother and Child

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Vesa Cosmin Mihai<sup>1</sup>, Vidican Madalina Ioana<sup>1</sup>, Petchesi Codruta<sup>2</sup>, Bembea Marius<sup>3</sup>, Jurca Claudia Maria<sup>3</sup>

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